Cystic Fibrosis

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What is Cystic Fibrosis?

Cystic fibrosis (CF) is one of the most common inherited diseases in US Caucasians.[1] It is caused by a failure of a protein that maintains the chloride (salt) balance in the body. This causes respiratory problems (breathing problems and lung infections), digestive problems (difficulty in absorbing some types of foods), and infertility.[1] CF does not cause mental retardation or birth defects.[1]

The symptoms of CF can be highly variable; they can be mild in some people and cause a life-threatening illness in others. With extensive treatment, the average person with CF lives into the third decade.[1]

Who is at risk for cystic fibrosis?

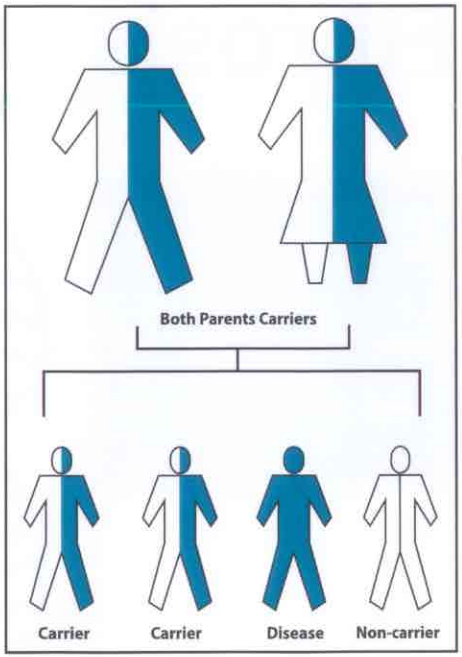
Cystic fibrosis is most common in Caucasians, but is found in all ethnic groups.[1] Even if an individual has no family history of CF or a child with CF, it is possible to be a CF carrier. Approximately 1 in 2500 Caucasians is born with the disease.[2] The carrier frequency of several ethnic groups is shown in Table 1. The chance of being a carrier is higher for those who have a family history of CF.

Table 1. Carrier frequency of CF

Ethnicity	Carrier Frequency
Caucasian (non-Hispanic)	1/25[2]
Ashkenazi Jewish	1/25[2]
Hispanic American	1/46[2]
African American	1/65[2]

What causes CF?

Cystic fibrosis is an inherited genetic condition.[1] It is inherited in a recessive manner. That means that both parents must be carriers to have an affected child. Being a carrier does not affect a person's health. Only when both parents are carriers of CF can the disease occur in their children. When both parents are carriers of CF, there is a 25% chance with each pregnancy that the child will have cystic fibrosis.[3]



How does the carrier test work?

Carrier testing is a special gene test. Everyone has genes. Genes are the blueprints that tell the body how to function. Genes are found in cells, such as blood cells or skin cells. If a gene has a problem, called a mutation, it does not work properly. CF carrier testing is a blood test that looks for mutations in the CF gene.

How accurate is the test?

There are many possible mutations in the CF gene. Some are rare and there may be some that have not been discovered yet. LabCorp tests for the most common CF mutations. A negative test significantly lowers the chance that a person is a carrier, but there is still a small chance that a rare mutation is present.

The following table shows the chance of being a carrier of CF when the LabCorp test is negative.[2] This table applies only to those people who do not have a family history of CF. For an individual with a family history and a negative CF test result, LabCorp will determine the specific risk of being a carrier of cystic fibrosis.

Ethnicity	Carrier detection rate for the 32 CF mutations	CF Carrier risk prior to testing	CF carrier risk after a negative result for 32 mutations
Ashkenazi Jewish	97%[2]	1/25[2]	1/800[2]
Caucasian (non-Hispanic)	90%[2]	1/25[2]	1/240[2]
Hispanic American	67-73%[4]	1/46[2]	1/146-1/167[5]
African American	69%[2]	1/65[2]	1/207[2]
Other	——	——	Insufficient data

What does it mean if the test is positive?

If the CF test indicates a person is a carrier, the next step is to test his or her partner. Both parents must be carriers to have an affected child. If the partner has a negative test result, the chance of having a baby with CF is less than 1/100. If the test is positive, the couple has a 25% chance with each pregnancy of having a child with CF.

What can a couple do if there is a risk of having affected children?

There are several options for couples who are at risk of having a child with cystic fibrosis. Many include personal choices that are best discussed in private by the couple and health care provider(s). A genetic counselor is a special type of health care provider who has expertise in genetic disorders such as CF, genetic testing, and the choices available to a couple during pregnancy. Two available options that may be discussed with a genetic counselor are CVS (chorionic villus sampling), a test done between 10 and 14 weeks of gestation, and amniocentesis, a test done between 15 and 20 weeks of gestation. Many other options are available and should be discussed with appropriate health care provider(s).

Why is CF testing recommended?

Cystic fibrosis is a common disorder for Caucasians. The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) have recommended that carrier screening be offered to all couples when at least one member of the couple is Caucasian and pregnant or considering pregnancy.[3] CF testing is not required; it is an option. Couples might choose to have carrier screening if prenatal diagnosis for CF is important to them, or if they want early diagnosis and treatment for their children at risk for CF. Couples of non-Caucasian ethnic groups may also request CF screening.[3] Choosing CF carrier screening is a personal choice that should be discussed in private with health care provider(s) and/or genetic counselor(s).

References

1. Brown T, Langfelder Schwind E. Update and review: Cystic fibrosis. *J Genet Couns.* 1999; 8(3):137-162.

2. Richards CS, Bradley LA, Amos J, et al. Standards and Guidelines for CFTR Mutation Testing. *Genet Med.* 2002; 4(5):379-391.

3. American College of Obstetricians and Gynecologists, American College of Medical Genetics. *Preconception and Prenatal Carrier Screening for Cystic Fibrosis: Clinical and Laboratory Guidelines.* Washington, DC: ACOG; Oct 2001.

4. This data is pooled from a, b, and c below.
   a. Richards CS, Bradley LA, Amos J, et al. Standards and Guidelines for CFTR Mutation Testing. *Genet Med.* 2002; 4(5): 379-391.
   b. Wang J, Bowman M, Hsu E, Wertz K, Wong L. A novel mutation in the CFTR gene correlates with severe clinical phenotype in seven Hispanic patients. *J Med Genet.* 2000; Mar; 37(3):215-218.
   c. Kharrazi M. CF patient registry [preliminary data]. Cited: 5 September 2002; www.dhs.cahwnet.gov/org/pcfh/gdb/cfwebsite/prelimres.htm.

5. Based on internal Bayesian calculation on file.

This publication and the information contained within are not intended to serve as a disclosure or informed consent nor to comply with any applicable laws or regulations. Your physician will provide the information about this test and any informed consent form necessary or required.

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